Parietal foramina in Saethre-Chotzen syndrome

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SUMMARY A father and son with Saethre-Chotzen syndrome and parietal foramina are described, to draw attention to this little known feature of the syndrome.

Saethre-Chotzen syndrome is characterised by craniosynostosis, facial asymmetry, low set frontal hairline, ptosis of the eyelids, deviated nasal septum, prominent crus of the ears, and a variable degree of brachydactyly and partial cutaneous syndactyly of the second and third fingers.^{1,2} Inheritance is autosomal dominant, mostly reported with a high degree of penetrance, although in a series reported by Carter *et al*³ there was evidence of incomplete penetrance. Expression is very variable. It is likely that many cases are undiagnosed because of lack of both serious cosmetic problems and medical complications.¹

Parietal foramina are defects in the parietal bone, usually located on each side of the posterior one-third of the sagittal suture.⁴ Small foramina, less than 1 mm in diameter are common and are said to occur in 60 to 70% of adults.⁵ Larger defects, of up to several centimetres in diameter, are less common and may occur as an isolated trait, either sporadically or autosomal dominantly inherited, or in association with various abnormalities.^{4,5} There is only one other report of an association of parietal foramina and Saethre-Chotzen syndrome.⁶

Case reports

CASE 1

This patient, a male, was born after a normal pregnancy to a 24 year old mother and a 33 year old father, who is case 2. Delivery was at term by caesarian section for breech presentation. The birth weight was 4.14 kg. The forehead was markedly protuberant with palpable thickening of the metopic suture and overlying soft tissue swelling (fig 1). The nasal bridge was markedly depressed. The skull was brachycephalic and a large midline bony defect of both parietal bones was palpable. There was a

FIG 1 Case 1 aged 3 days.

small beaked nose, prominent crus of the ears, small chin, hairy forehead, and hypotelorism (figs 2, 3). The hands and feet were normal.

A skull x-ray showed synostosis of the coronal sutures and of the lower part of the metopic suture. The margins of the upper part of the metopic suture were everted and continuous with large symmetrical defects in the parietal bones which extended to the midline (fig 4). In the region of the posterior part of the sagittal suture were small ossified segments of bone. The lambdoid sutures were normal. The orbits were small with hypotelorism. A CT brain scan and lymphocyte chromosome karyotype were normal.

There were no neonatal problems and the child was admitted at 5 months of age for frontal

FIG 2 Case 1 aged 3 days.

FIG 3 Case 1 aged 3 days.

cranioplasty and supraorbital ridge advancement. Developmental progress at 10 months of age was normal.

CASE 2

The father of case 1 was born in Ireland and reported having had such a 'soft head' that he had been cared for in a nursing home for the first 2 years of life. In the past he had had a cleft palate repaired, four operations for congenital right ptosis which had

FIG 4 Skull x-ray, case 1.

persisted, an operation for deviated nasal septum at 15 years, and treatment for occasional seizures until 19 years. He reported being the shortest of the family at 163 cm. His mother he reported as 168 cm and his father 180 cm tall. He seemed of normal intellect.

On examination, there was brachycephaly, facial asymmetry, low frontal hairline, supraorbital recession, beaked nose, maxillary hypoplasia, right ptosis, notched upper lip, repaired cleft palate, and palpable posterior parietal bone defects bilaterally (figs 5, 6). The hands and feet were normal. A recent skull x-ray showed no visible sutures and brachycephaly, consistent with previous premature synostosis of at least the coronal sutures. Large bilateral posterior parietal foramina were present (fig 7).

Discussion

Large parietal foramina are thought to arise from a midline ossification defect which involves both parietal bones and which is present at birth. Small areas of bone formation appear in the defect parasagittally and gradually form a midline sagittal strip of bone which divides the parietal defect into two foramina.⁵

Presumably the father's 'soft head' at birth relates to a large parietal bone defect from which the parietal foramina arose. Similarly, the son's large midline parietal bone defect is likely to be the forerunner of parietal foramina.

FIG 5 Case 2, father of case 1.

FIG 7 Skull x-ray, case 2.

FIG 6 Case 2.

Parietal defects have been described in a variety of other cases of craniosynostosis including a 'Crouzon-variant',⁷ an unusual form of acrocephalosyndactyly,⁸ an adult male with oxycephaly and Klippel-Feil deformity,⁹ and in monozygotic twins with 'craniofacial dysostosis'.¹⁰ Parietal

foramina occur with other skeletal defects, such as faulty ossification of the clavicles.¹¹

Only one other report of the association of parietal foramina and Saethre-Chotzen syndrome was found, in which Friedman *et al*⁶ described a kindred of 15 persons with Saethre-Chotzen syndrome, of which three of six x-rayed had parietal foramina.

Parietal foramina may reflect a general abnormality of skull bone formation in Saethre-Chotzen syndrome, which is characterised by an unevenness of the rate of ossification, resulting in premature craniosynostosis of some areas and delayed ossification of others, and perhaps accounting for facial asymmetry. It remains to be seen if parietal foramina are more common in Saethre-Chotzen syndrome than in other forms of craniosynostosis.

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